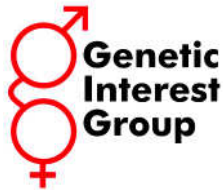


Acknowledgements.

We would like to thank all those who took part in the focus groups, interviews, and on-line survey and all who helped in the development of this guide.

The *Family Route Maps* project is funded by the charity Jeans for Genes and an unrestricted educational grant from Genzyme Therapeutics Ltd.



Genetic Interest Group (GIG)

Unit 4D, Leroy House

436 Essex Road, London, N1 3QP

Tel: 0207 7043141

www.gig.org.uk

Registered Charity No 1114195

Company Registered by Guarantee 05772999

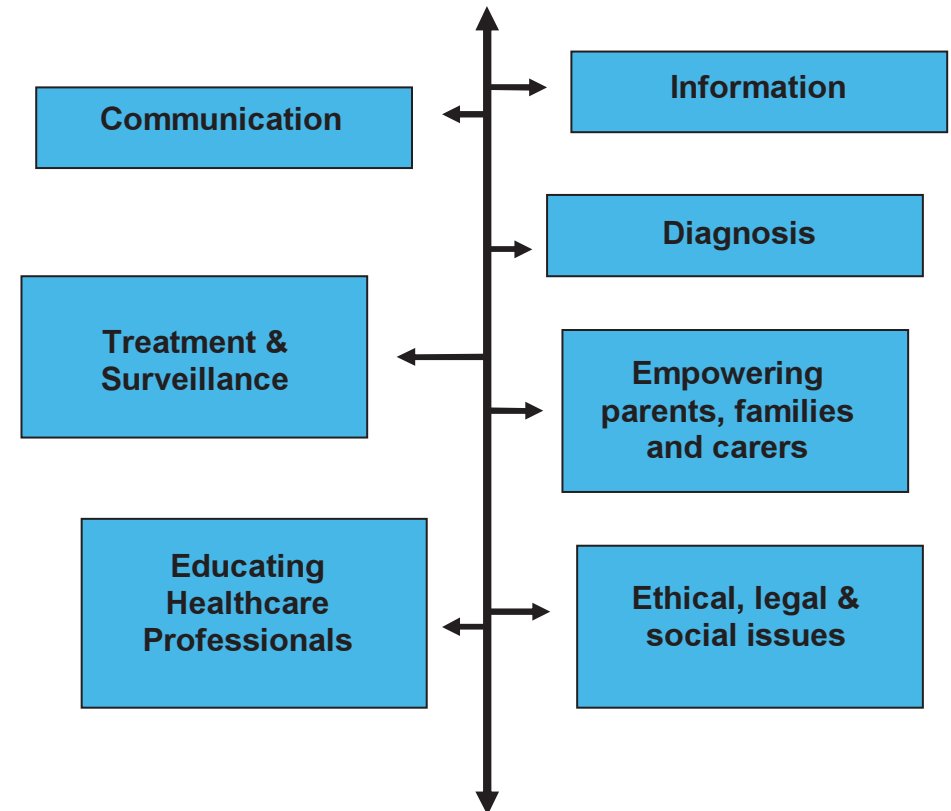
This leaflet has been prepared in good faith to provide patients with a guide to current services and information. Neither GIG nor SWAN can be held responsible for the accuracy of the information it contains. Links to other organisations are included for information purposes only and are not recommendations from GIG or SWAN.

Date published: February 2008

Date for review: February 2009

SWAN (Syndromes Without A Name) *Family Route Map*

This Family Route Map is a guide to current services and information.



Registered Charity Number 1074829

INTRODUCTION

This leaflet is designed to provide signposts to sources of current information and appropriate services for patients, their families and carers, together with healthcare professionals. The issues and concerns raised during a series of patient focus groups in 2006 organised by the Genetic Interest Group (GIG) identified seven themes (see front cover) which have been used as the basis for developing this resource with the help of patients, families and carers, and clinical staff with experience and expertise of this condition.

SWAN

The charity SWAN (Syndromes Without A Name)

- Provides support and information on a variety of subjects.
- Provides a listening ear through the helpline.
- Links families together for mutual support and exchange of information.
- Campaigns on behalf of families who have a child with an undiagnosed condition.

Their Mission Statement:

To preserve and protect the health and promote the welfare of children who suffer from undiagnosed conditions.

SWAN will be pleased to provide information and try to answer your queries. If you have a child with an undiagnosed condition or are a professional working with children with un-named syndromes and you would like to become a member of SWAN.

The charity produce a regular Newsletter containing useful advice and featuring letters from parents of other children without a diagnosis.

Please telephone their helpline on 01922 701234 or email info@undiagnosed.org.uk

SWAN has a website www.undiagnosed.org.uk

Links to other useful organisations:

BIBIC (British Institute for Brain Injured Children) www.bibic.org.uk
Tel: 01278 684060

Birth Defects Foundation www.bdfnewlife.co.uk Tel: 08700 707020

British Society for Human Genetics (BSHG): www.bshg.org.uk

Carers UK www.carersuk.org Tel: 0808 808 7777

Contact-A-Family: www.cafamily.org.uk Tel: 020 7608 8700

Department of Health have produced a set of 'Questions to ask' to take with you to your appointment with a specialist. Available in several different languages; use the Search option on their website. www.dh.gov.uk

Directgov www.direct.gov.uk A website with help on benefits and entitlements

Disability Benefits Office Tel: 08457 123456

Disablement Income Group Scotland www.digscotland.org.uk
Tel: 0131 555 2811

Early Support have a website www.earlysupport.org.uk Tel: 0845 602 2260

GIG (Genetic Interest Group) have a range of patient leaflets on their website www.gig.org.uk/eurogentest_patientleaflets.htm Tel: 020 7704 3141

HEADWAY (The Brain Injury Association) www.headway.org.uk
Tel: 0808 800 2244

National Institute of Conductive Education www.conductive-education.org.uk

The Challenging Behaviour Foundation www.challengingbehaviour.org.uk
Tel: 01634 838739

The Family Fund www.familyfund.org.uk Tel: 0845 1304542

Special Kids in the UK www.specialkidsintheuk.org are a charity providing information, support and contact between families of children with special needs.
Address: PO Box 617, Addlestone, KT15 9AP

Transition Information Network www.transitioninforonetwork.org.uk aim to improve the experience of disabled young people's transition to adulthood.

Wellchild www.wellchild.org.uk The charity has a Helpline, Tel: 0808 801 0330

Whizz Kidz www.whizz-kidz.org.uk can provide help to fund equipment. This charity also have a Family Advice Worker Tel: 020 7798 6114

OTHER INFORMATION

Insurance

It is common to have difficulties finding adequate and affordable insurance policies once you have a pre-existing condition.

The following company offers travel insurance for people with pre-existing conditions. There may be others available and this should not be taken as a recommendation:

- The insurance group Banner - see www.bannergroup.com have agreed to look at and consider insuring anyone, however they cannot guarantee that they will cover everyone for all conditions, it will have to be on an individual basis.

Financial help

CAB (Citizens Advice Bureau) can help with advice locally about benefits and can be found through your phone directory.

Psychological help

Psychological counselling is not commonly offered on diagnosis or even later on. If you feel that you need extra help in coming to terms with your condition or any other aspect of your life, don't be afraid to ask for this via your GP.

Physiotherapy

Many parents said that physiotherapy had been extremely helpful but may be limited to only a short course. You can ask your GP or Community Paediatrician for a referral.

Education

If you have concerns about your child requiring extra support with learning or if they are experiencing other problems at school discuss this with your Community Paediatrician or ask for a referral to specialist services from your GP or Health Visitor.

A leaflet entitled 'Statutory assessment—Education' (leaflet code ES26) from *Early Support* explains the process of requesting an assessment for your child and provides examples of letters to help you. It also describes what a **Statement of Special Educational Needs** is. This can be obtained from the *Early Support* website www.earlysupport.org.uk or by phoning 0845 602 2260.

INFORMATION

A syndrome is a collection of symptoms, often appearing in combination with one another. Children may have combinations of various conditions including;

- Epilepsy
- Feeding problems
- Poor mobility
- Unexplained pain
- Speech problems
- Neurological problems
- Heart problems
- Skin problems
- Growth problems
- Developmental delay
- Vision problems
- Genetic conditions
- Behavioural problems
- Orthopaedic problems
- Emotional difficulties
- Hearing problems

Genetic Testing:

Genetic testing and counselling is available at 23 regional NHS genetic centres throughout the UK; please see the website of www.bshg.org.uk for a list and also in their section 'For Patients' there is an explanation of what Genetic Counselling is and is not. A referral to a genetic centre is usually made through your GP or specialist. Importantly, if you were seen several years ago in a Clinical Genetics Unit you may wish to be reviewed and in this case it might be possible to contact them directly yourself.

There are a number of issues surrounding genetic testing particularly in relation to children and as such, many patients may wish to be seen and counselled by a consultant clinical geneticist as early on as possible. The Genetic Interest Group (GIG) website has a series of leaflets explaining more about inherited conditions and includes; a glossary of terms used in genetics, and useful questions to ask when going for an appointment.

Please see www.gig.org.uk/eurogentest_patientleaflets.htm

The UKGTN (UK Genetic Testing Network) has produced a patient leaflet to help understand testing. Available at their website www.ukgtn.nhs.uk/gtn/UKGTN-information/Patient-leaflet.html

DIAGNOSIS, TREATMENT AND SURVEILLANCE

Although estimates vary enormously the most commonly quoted figure suggests that between 30% - 40% of special needs children do not have a specific diagnosis. Non diagnosis means more difficulty in claiming benefits and accessing support services.

The charity, **Contact A Family**, have produced a leaflet for parents with undiagnosed children. It is available from their website www.cafamily.org.uk/undiagno.html or phone their helpline 0808 808 3555 for a copy.

The Early Support unit also have a leaflet 'When your child has no diagnosis' (leaflet code ES16) which can be obtained from their website www.earlysupport.org.uk or by phoning 0845 602 2260.

Personal Passport

Some parents have found it useful to create a 'Personal Passport' for their child which accompanies them when outside of the home. It contains medical and other information, for example, if your child communicates using speech or in another way, if your child prefers not to be touched, or what sorts of things they don't like to eat etc. The medical information can be helpful in emergencies, for example, it might list drugs that your child should not be given plus the name of your child's hospital consultant to contact for further information.

TIP

MedicAlert Foundation. MedicAlert ID emblems enable emergency medical staff to call a 24-hour helpline number to obtain detailed information on your child's medical condition, a summary of which is engraved on the emblem. Details can be found on the website at www.medicalert.org.uk

Second Opinion

In the UK, patients have the right to ask for a second opinion via their first specialist or through their GP. They are not, however, entitled to receive that second opinion if the clinicians do not think it necessary. A patient may appeal against any decision via their Primary Care Trust (PCT).

For more information on the Referral Process please see the GIG website: www.gig.org.uk/docs/referrals.pdf

Hospital Appointments

Your child may have appointments with many different hospital specialties. Discuss with your Community Paediatrician if they can help schedule your appointments and make referrals in the early years. Parents may request a multi-disciplinary team meeting to discuss their children's complex needs. Remember to take a pencil and paper with you to appointments and write down anything you are unsure of as many people say they cannot remember when they get home.

TIP

Say how you feel: if you don't want medical students participating in your appointments; or clinical staff discussing your child in front of them, don't be afraid to say so.

INFORMATION FOR HEALTHCARE PROFESSIONALS

Information and articles about SWAN are available from the SWAN website www.undiagnosed.org.uk

More general information can be found on the following websites;

The National Organization for Rare Diseases
www.rarediseases.org

Orphanet (European database) www.orpha.net
Free-access website providing information on rare diseases.

The NHS National Genetics Education and Development Centre
www.geneticseducation.nhs.uk
provide information and resources for healthcare professionals.

Genetics in Family Medicine: The Australian Handbook for General Practitioners www.gpgenetics.edu.au